

L5 ANSWER 1 OF 1 CAPLUS COPYRIGHT 2002 ACS
 AN 1996:355569 CAPLUS
 DN 125:50211
 TI Mutational scanning of large genes by extensive PCR multiplexing and two-dimensional electrophoresis: application to the RB1 gene
 AU Van Orsouw, Nathalie J.; Li, Daizong; van der Vlies, Pieter; Scheffer, Hans; Eng, Charis; Buys, Charles H. C. M.; Li, Frederick P.; Vijg, Jan
 CS Mol. Genet. Section, Beth Israel Hosp. and Harvard Med. Sch., Boston, MA, 02215, USA
 SO Hum. Mol. Genet. (1996), 5(6), 755-761
 CODEN: HMGEES; ISSN: 0964-6906
 DT Journal
 LA English
 AB With the rapid increase in the no. of identified human disease genes, the development of accurate and cost-efficient mutation tests has become opportune. Here we present a combination of extensive PCR multiplexing and two-dimensional (2-D) DNA electrophoresis to screen for mutations in 26 exons of the retinoblastoma (RB1) tumor suppressor gene. In 2-D electrophoresis, fragments are sepd. according to size and base pair sequence in non-denaturing and denaturing gradient gels, resp. All target fragments, designed to have optimal melting characteristics, were prepd. in a two-step PCR (a 6-plex long-PCR pre-amplification and a subsequent 25-plex short-PCR) followed by heteroduplexing. The mixt. of PCR amplicons was then subjected to 2-D electrophoresis under a single set of exptl. conditions. With this design, 35 previously identified mutations in 18 different exons were detected in 33 bilateral retinoblastoma patients. These results suggest that 2-D electrophoresis in this format provides a generally applicable, practical and fast way to diagnose with high accuracy large genes for a broad spectrum of possible disease-causing mutations.

=> d hitseq

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 IT **178305-13-0**
 FL: BUU (Biological use, unclassified); PRP (Properties); THU (Therapeutic use); BIOL (Biological study); USES (Uses)
 (PCR primer; mutational scanning of large genes by extensive PCR multiplexing and two-dimensional electrophoresis and application to the RB1 gene)
 RN 178305-13-0 CAPLUS
 CN DNA, d(C-G-C-C-C-G-C-C-G-C-G-C-C-C-C-G-C-G-C-C-C-G-T-C-C-C-G-C-C-C-G-A-C-A-T-G-T-A-A-A-G-G-A-T-A-A-T-T-G-T) (PCI) (CA INDEX NAME)
 NTE singlestranded
 SEQ 1 cgcctgcgcgc gcccgcgcgc cgtccgcgc gacatgtaaa ggataattgt

=>

=> d his

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78798 S CGACATGT/'SQSN

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5304 S L1
L3 0 S L2 AND SQL<50

L4 FILE 'REGISTRY' ENTERED AT 09:17:26 ON 18 APR 2002
4 S CGACATGT/'SQSN AND 50/SQL

L5 FILE 'CAPLUS' ENTERED AT 09:18:03 ON 18 APR 2002
1 S L4

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